

Health Care Provider Fact Sheet

Disease Name

Congenital Adrenal Hyperplasia

Acronym

CAH

Disease Classification

Endocrine Disorder

Symptom onset

INFANTS WITH CAH DO NOT APPEAR ILL AT BIRTH, BUT MAY, WITHIN THE FIRST FEW WEEKS OF LIFE, EXPERIENCE A SALT-LOSING CRISIS WHICH CAN LEAD TO SERIOUS ILLNESS AND DEATH.

Symptoms

Congenital adrenal hyperplasia (CAH) results from a deficiency in one or another of the enzymes of cortisol biosynthesis. In about 95% of cases, 21-hydroxylation is impaired in the zona fasciculata of the adrenal cortex so that 17-hydroxyprogesterone (17-OHP) is not converted to 11-deoxycortisol. Because of defective cortisol synthesis, ACTH levels increase, resulting in overproduction and accumulation of cortisol precursors, particularly 17-OHP, proximal to the block. This causes excessive production of androgens, resulting in virilization.

Natural history without treatment

If untreated, children with CAH will experience abnormally rapid growth early in childhood (but stunted in the long run) and early appearance of body hair. Babies with the salt-wasting form of CAH (about 75 percent of cases) are at risk for rapid, uncontrolled loss of salt from the body that can result in death. The imbalance of hormones before birth may cause some girls to have ambiguous genitalia.

Treatment

Daily supplements of the hormone cortisol, and in many cases a salt-retaining hormone. To prevent problems, treatment must begin shortly after birth.

Physical phenotype

Ambiguous genitalia in females

Inheritance

Autosomal recessive

General population incidence

1 in 21,500

OMIM Link

<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=201910>

Genetests Link

www.genetests.org

Support Group

Congenital Adrenal Hyperplasia Research Education & Support
<http://www.caresfoundation.org>

MAGIC Foundation for Children's growth (MAGIC)
<http://www.magicfoundation.org>

National Organization for Rare Diseases
<http://www.rarediseases.org>